

THE BRAIN IN CHARGE SYNDROME: FOR THE NEUROLOGIST

Bruce R. Korf, M.D., Ph.D.

Medical Director, Harvard-Partners Center for Genetics and Genomics

77 Ave Louis Pasteur, Suite 642, Boston, MA 02115

bkorf@partners.org (617) 525-5750

Brain anomalies seen in CHARGE

Cranial nerve anomalies (70-90%)

I - olfactory - (arhinencephaly in 42%)

II - optic (colobomas in 80%)

III, IV, V, VI rarely affected

VII – facial palsy, UL or BL (43%)

VIII - acoustic (60 - 80%+)

IX, X glossopharyngeal/vagus (50%+)

XI accessory - rarely affected

XII hypoglossal - rarely affected

Structural anomalies (less common)

agenesis of the corpus callosum

arhinencephaly

holoprosencephaly

hydrocephalus

cerebral dysgenesis

Mondini defect

hindbrain defects

 cerebellar hypoplasia

 Dandy-Walker malformation

Other

 seizures

 behavior abnormalities

 learning disabilities

Development: although it has been reported in the past that a majority of children with CHARGE are mentally retarded, this is not necessarily the case. It is extremely important for parents to realize that their child may have significant (even near-normal) potential, given accurate diagnosis and appropriate management of sensory deficits and other complications of CHARGE.

Diagnostic tests/referrals

 MRI of brain

 CT

 EEG (if suspect seizures)

 Evoked potentials (vision/hearing)

 Other hearing tests (otolaryngology)

 Dilated eye exam (ophthalmology)

 Developmental testing (developmental pediatrics/deafblind specialist)

Medical treatment caveats

Possible unexpected reaction to anesthesia: Some children with CHARGE are resistant to sedation, while others are slow to recover from anesthesia. Because of risks of anesthesia, it may be appropriate to combine surgical procedures in these children.

Laryngomalacia or tracheomalacia is common in CHARGE and can result in surgical or anesthetic complications. The mortality rate in children with this combination of features is high.

Reflux, facial palsy and other cranial nerve anomalies may additionally compromise feeding in these children. There is a very high risk for repeated aspiration pneumonia.

If MRI is being considered, also consider a few more cuts to view the inner and middle ear, as there is a very high frequency of ossicular malformations and/or Mondini defect.

Non-medical management:

Cranial nerve IX, X, and VII anomalies can have an enormous impact on feeding. Feeding issues and common aspiration pneumonia are among the most important day-to-day issues for families. Often a feeding specialist can help the family cope with these issues.

References

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THE BRAIN IN CHARGE: PARENT INFORMATION

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bkorf@partners.org (617) 525-5750

and

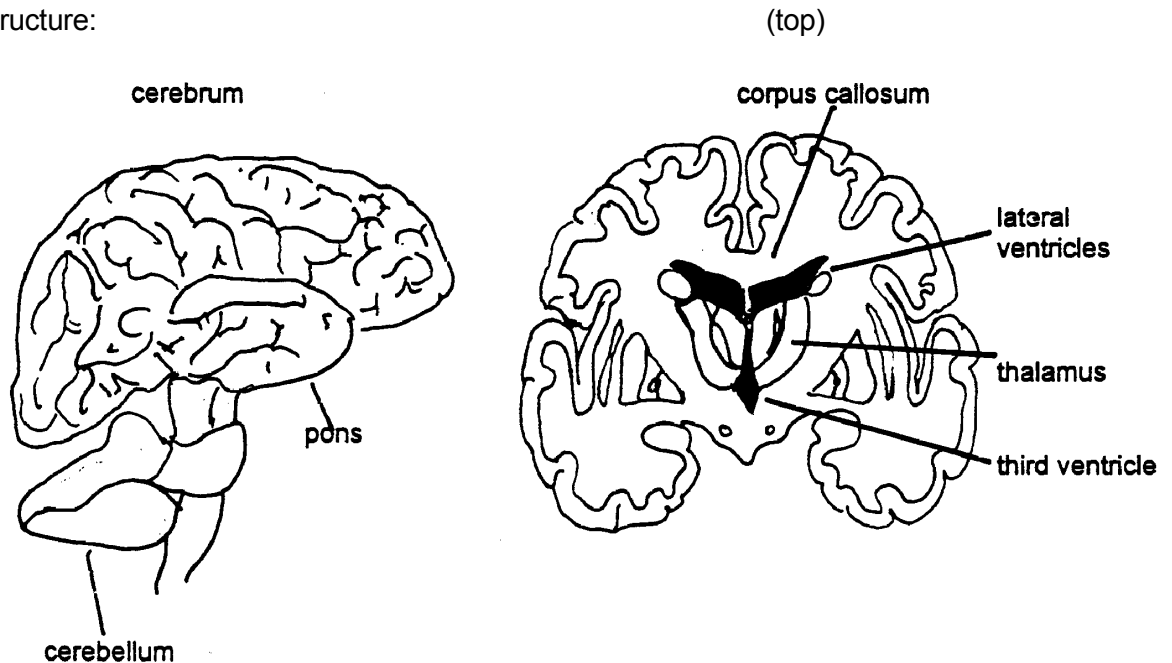
Meg Hefner, M.S.

Assistant Professor of Pediatrics, Division of Medical Genetics, St. Louis University School of Medicine, 1465 S. Grand Blvd. St. Louis, MO 63104

Hefnerma@slu.edu or Meg@chargesyndrome.org (314)768-8730

NORMAL BRAIN STRUCTURE AND FUNCTION:

Brain structure:



Cranial nerves: there are 12 cranial nerves, which begin in the brain and extend to structures in the head and neck. These nerves provide both motor control and sensation and include nerves involved in the senses of smell, taste, hearing, and vision, as well as movements of the eyes, face, tongue, palate, and swallowing. Some cranial nerves are also involved in the control of heart rate and movements of the gastrointestinal tract. Cranial nerve anomalies are very frequent in CHARGE.

BRAIN ABNORMALITIES SEEN IN CHARGE

Cranial Nerve Anomalies:

Anomaly	Frequency	Tests Used	Specialist
I – olfactory (smell)	? 40%	Clinical assessment	neurologist/otolaryngologist
II – optic nerve Coloboma	80-90%	Dilated exam Visual evoked responses	Ophthalmologist
VII – facial palsy	42%, usually one sided	Clinical assessment	neurologist
VIII – sensorineural hearing loss	60-80%	ABR Other hearing tests	pediatric audiologist otolaryngologist
IX, X swallowing and oromotor problems	50%+	Barium swallow Laryngoscopy	otolaryngologist OT, PT, speech pathology
XII – tongue	rare	Clinical assessment	neurologist

Structural brain anomalies:

A variety of structural malformations of the brain have been reported in children with CHARGE. Pretty much any brain anomaly is consistent with CHARGE; none are extremely common. A neurologist may order brain imaging such as MRI or CT scan to look for possible structural brain anomalies. Because not all children have had imaging performed, it is not possible to list the frequency of each anomaly.

1. Arhinencephaly: absence of the olfactory lobes
2. Holoprosencephaly: abnormal formation of the cerebral hemispheres.
3. Agenesis of the corpus callosum: lack of fiber tract connecting the two hemispheres.
4. Cerebral dysgenesis: abnormal formation of the cerebral cortex.
5. Hydrocephalus: increased fluid in the ventricles of the brain

Other brain abnormalities described in CHARGE

1. Seizures: diagnosed by EEG
2. Behavior abnormalities: See DEVELOPMENT sections
3. Learning disabilities: SEE Development section

EFFECT OF ABNORMALITY ON CHILD

Cranial nerve abnormalities: cannot be surgically corrected. Only a few (hearing loss, swallowing problems) are amenable to management.

Olfactory nerve (I):

This nerve controls the sense of smell. If it is abnormal, the child's sense of smell (and therefore taste) will be absent or abnormal. This can complicate feeding problems.

Optic nerve (II):

A majority of children with CHARGE have colobomas. Optic nerve colobomas can severely affect vision. See EYE section.

Facial palsy (VII):

Droopiness or weakness of one (unilateral, UL) or both sides (bilateral, BL) of the face. This may affect feeding, with drooling on the affected side. If bilateral, there may be problems keeping food in the mouth. Facial palsy can have an effect on facial expression. In some cases, the eye on the affected side may not close completely and need to be kept moist with artificial tears. In some cases, facial nerve palsies get better over time. See FEEDING section.

Acoustic nerve (VIII):

Abnormalities of cranial nerve VIII will result in sensorineural hearing loss (see EARS AND HEARING).

Glossopharyngeal and vagus nerves (IX and X):

These nerves are involved in coordination of suck and swallow and with some of the mouth movements involved in speech. Feeding and swallowing problems are very common in children with CHARGE (see FEEDING). Many of these problems get better over time (often years), due to a combination of maturation of the nerve and use of appropriate therapies.

Structural brain abnormalities: most are not treatable

1. Arhinencephaly will cause lack of smell. This can be difficult to diagnose, but has been described very often at autopsy in CHARGE.
2. Holoprosencephaly is a serious brain abnormality, which is likely to result in significant mental retardation.
3. Agenesis of the corpus callosum is often (but not always) associated with learning disabilities and/or mental retardation.
4. Hydrocephalus can be associated with learning disabilities or mental retardation. Hydrocephalus is treated by placing a shunt.
5. Cerebral dysgenesis: many children with CHARGE have structural brain abnormalities noted on MRI or CT scan. The significance in terms of functioning is not always clear.

Other brain abnormalities described in CHARGE

1. Seizures: diagnosed by EEG, often effectively treated with medication
2. Behavior abnormalities: See DEVELOPMENT sections.